# Your Guide to Understanding Genetic Conditions

# STK11 gene

serine/threonine kinase 11

### **Normal Function**

The *STK11* gene (also called *LKB1*) provides instructions for making an enzyme called serine/threonine kinase 11. This enzyme is a tumor suppressor, which means that it helps keep cells from growing and dividing too fast or in an uncontrolled way. This enzyme helps certain types of cells correctly orient themselves within tissues (polarization) and assists in determining the amount of energy a cell uses. This kinase also promotes a type of programmed cell death known as apoptosis. In addition to its role as a tumor suppressor, serine/threonine kinase 11 function appears to be required for normal development before birth.

# **Health Conditions Related to Genetic Changes**

### breast cancer

Inherited changes in the *STK11* gene greatly increase the risk of developing breast cancer, as well as other types of cancer, as part of Peutz-Jeghers syndrome (described above). These mutations are thought to account for only a small fraction of all breast cancer cases.

### ovarian cancer

# Peutz-Jeghers syndrome

Inherited mutations in the *STK11* gene cause Peutz-Jeghers syndrome, a condition characterized by the development of noncancerous growths called hamartomatous polyps in the gastrointestinal tract and a greatly increased risk of developing several types of cancer. More than 340 *STK11* gene mutations have been identified in people with this condition. Many of these mutations result in the production of an abnormally short, nonfunctional version of the serine/threonine kinase 11 enzyme. Other mutations change single protein building blocks (amino acids) used to build the enzyme. Mutations in the *STK11* gene impair the enzyme's tumor suppressor function, allowing cells to grow and divide without control or order. This uncontrolled cell growth can lead to the formation of hamartomatous polyps and cancerous tumors.

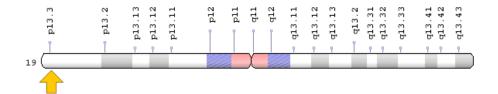
#### other cancers

Noninherited (somatic) mutations in the *STK11* gene have been found in various forms of cancer. Somatic mutations are acquired during a person's lifetime and are present only in certain cells. They do not occur as part of a cancer syndrome. Somatic *STK11* gene mutations have been identified in a form of lung cancer called non-small cell lung carcinoma, cervical cancer, colorectal cancer, an aggressive type of skin cancer called melanoma, and pancreatic cancer. These mutations impair the function of serine/threonine kinase 11, which can allow cells to grow and divide uncontrollably and contribute to the formation of a cancerous tumor.

# **Chromosomal Location**

Cytogenetic Location: 19p13.3, which is the short (p) arm of chromosome 19 at position 13.3

Molecular Location: base pairs 1,205,799 to 1,228,435 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

# Other Names for This Gene

- LKB1
- PJS
- serine/threonine kinase 11 (Peutz-Jeghers syndrome)
- Serine/threonine-protein kinase 11
- STK11\_HUMAN

#### Additional Information & Resources

#### **Educational Resources**

- Cancer Syndromes (2009): Peutz-Jeghers Syndrome https://www.ncbi.nlm.nih.gov/books/NBK1826/
- National Cancer Institute: Genetics of Breast and Gynecologic Cancers (PDQ) https://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq

# GeneReviews

 Peutz-Jeghers Syndrome https://www.ncbi.nlm.nih.gov/books/NBK1266

# Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28STK11%5BTIAB%5D%29+OR+%28serine/threonine+kinase+11%5BTIAB%5D%29%29+OR+%28LKB1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

### **OMIM**

 SERINE/THREONINE PROTEIN KINASE 11 http://omim.org/entry/602216

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/STK11ID292.html
- Cancer Genetics Web: STK11 http://www.cancerindex.org/geneweb/STK11.htm
- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=STK11%5Bgene%5D
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene\_symbol\_report?q=data/ hgnc data.php&hgnc id=11389
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/6794
- UniProt http://www.uniprot.org/uniprot/Q15831

# Sources for This Summary

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